immune thrombocytopenia

Immune thrombocytopenia is a disorder characterized by a blood abnormality called thrombocytopenia, which is a shortage of blood cells called platelets that are involved in clotting.

Affected individuals can develop frequent bruising or red or purple spots (purpura) on the skin caused by bleeding just under the skin's surface. People with immune thrombocytopenia can have significant bleeding episodes, such as nose bleeds (epistaxis) or bleeding in the moist lining (mucosae) of the mouth. In severe cases, individuals may have gastrointestinal bleeding or heavy or prolonged menstrual bleeding (menorrhagia). In very rare instances, bleeding inside the skull (intracranial hemorrhage) can occur, which can be life-threatening. A greater reduction in platelet numbers is often associated with more frequent bleeding episodes and an increased risk of severe bleeding.

While immune thrombocytopenia can be diagnosed at any time, there are two periods when the condition is most likely to develop: early childhood and late adulthood. In children, the reduction in platelets is usually sudden, but platelet levels usually return to normal levels within weeks to months. Immune thrombocytopenia in children is often preceded by a minor infection, such as an upper respiratory infection, but the relationship between the infection and immune thrombocytopenia is not clear. In adults, the development of immune thrombocytopenia is gradual and the condition tends to persist throughout life.

Frequency

The incidence of immune thrombocytopenia is approximately 4 per 100,000 children and 3 per 100,000 adults. It is likely that this condition is underdiagnosed because those with mild signs and symptoms often do not seek medical attention.

Genetic Changes

The genetic cause of immune thrombocytopenia is unclear. This condition occurs when the body's own immune system abnormally destroys platelets and makes fewer platelets than normal. Normally, the immune system produces proteins called antibodies, which attach to specific foreign particles and germs, marking them for destruction. People with immune thrombocytopenia produce antibodies that instead attack normal platelets. This destruction of platelets results in a shortage of these cells in affected individuals. Some of these antibodies also affect the cells that produce platelets (known as megakaryocytes), which leads to a decrease in platelet production, further reducing the number of platelets in the blood.

In some people with immune thrombocytopenia, the abnormal immune reactions may coincide with an infection by certain viruses or bacteria. Exposure to these foreign invaders may trigger the body to fight the infection, but the immune system also mistakenly attacks platelets.

Genetic variations (polymorphisms) in a few genes have been found in people with immune thrombocytopenia and may increase the risk of abnormal immune reactions. However, the contribution of these genetic changes to the development of immune thrombocytopenia is unclear.

When the condition is due to the targeted destruction of platelets by the body's own immune cells, it is known as primary immune thrombocytopenia. Immune thrombocytopenia following bacterial or viral infection is considered primary because the infection triggers a platelet-specific immune reaction, typically without any other signs or symptoms. However, immune thrombocytopenia can be a feature of other disorders, including autoimmune disorders such as systemic lupus erythematosus, which occur when the immune system malfunctions and attacks multiple tissues and organs in the body. Immune thrombocytopenia can also occur with blood disorders, including a form of cancer of the blood-forming tissue known as chronic lymphocytic leukemia, and human immunodeficiency virus (HIV) infection. In these cases, the condition is known as secondary immune thrombocytopenia.

Inheritance Pattern

Immune thrombocytopenia and other autoimmune disorders can run in families, but the inheritance pattern is usually unknown. People with a first-degree relative (such as a parent or sibling) with immune thrombocytopenia likely have an increased risk of developing the disorder themselves.

Other Names for This Condition

- autoimmune thrombocytopenia
- autoimmune thrombocytopenic purpura
- idiopathic thrombocytopenic purpura
- immune thrombocytopenic purpura
- ITP
- Werlhof disease

Diagnosis & Management

These resources address the diagnosis or management of immune thrombocytopenia:

- Genetic Testing Registry: Idiopathic thrombocytopenic purpura
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0043117/
- Johns Hopkins Medicine
  http://www.hopkinsmedicine.org/healthlibrary/conditions/hematology_and_blood_disorders/immune_thrombocytopenic_purpura_85,P00096/
- MedlinePlus Encyclopedia: Idiopathic Thrombocytopenic Purpura (ITP)
  https://medlineplus.gov/ency/article/000535.htm
- Seattle Children's Hospital
  http://www.seattlechildrens.org/medical-conditions/heart-blood-conditions/itp-symptoms/

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Idiopathic Thrombocytopenic Purpura (ITP)
  https://medlineplus.gov/ency/article/000535.htm
- Encyclopedia: Purpura
  https://medlineplus.gov/ency/article/003232.htm
- Health Topic: Platelet Disorders
  https://medlineplus.gov/plateletdisorders.html

Genetic and Rare Diseases Information Center

- Idiopathic thrombocytopenic purpura
  https://rarediseases.info.nih.gov/diseases/5194/idiopathic-thrombocytopenic-purpura

Additional NIH Resources

- National Heart, Lung, and Blood Institute
  https://www.nhlbi.nih.gov/health/health-topics/topics/itp

Educational Resources

- Ann & Robert H. Lurie Children's Hospital of Chicago
  https://www.luriechildrens.org/en-us/care-services/conditions-treatments/immune-thrombocytopenia-pupura/Pages/index.aspx
- Cincinnati Children's Hospital
  https://www.cincinnatichildrens.org/health/i/itp
- Cleveland Clinic
  http://my.clevelandclinic.org/health/articles/immune-thrombocytopenia
- Dana-Farber Cancer and Blood Disorders Center at Boston Children's Hospital
  http://www.danafarberbostonchildrens.org/conditions/blood-disorders/immune-thrombocytopenic-purpura.aspx
- Disease InfoSearch: Immune Thrombocytopenic Purpura
  http://www.diseaseinfosearch.org/Immune+Thrombocytopenic+Purpura/3775
- MalaCards: thrombocytopenic purpura, autoimmune
  http://www.malacards.org/card/thrombocytopenic_purpura_autoimmune
- Merck Manual Consumer Version
  http://www.merckmanuals.com/home/blood-disorders/platelet-disorders/immune-thrombocytopenia
- Orphanet: Immune thrombocytopenic purpura
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3002
- The Children's Hospital of Philadelphia
  http://www.chop.edu/conditions-diseases/idiopathic-thrombocytopenic-purpura-itp#.V6DoqaIgGKs

Patient Support and Advocacy Resources

- Contact a Family (UK)
  http://www.cafamily.org.uk/medical-information/conditions/i/immune-thrombocytopenia/
- Foundation for Women & Girls with Blood Disorders
  http://www.fwgbd.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/immune-thrombocytopenia/
- Platelet Disorder Support Association
  https://www.pdsa.org/
- The ITP Support Association (UK)
  http://www.itpsupport.org.uk/

Genetic Testing Registry

- Idiopathic thrombocytopenic purpura
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0043117/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22immune+thrombocytopenia%22+OR+%22ITP%22+OR+%22idiopathic+thrombocytopenic+purpura%22+OR+%22immune+thrombocytopenic+purpura%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Purpura,+Thrombocytopenic,+Idiopathic%5BMAJR%5D%29+AND+%28immune+thrombocytopenia%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- THROMBOCYTOPENIC PURPURA, AUTOIMMUNE
  http://omim.org/entry/188030

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/immune-thrombocytopenia

Reviewed: August 2016
Published: February 7, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services